



## REEP1 gene

receptor accessory protein 1

### Normal Function

The REEP1 gene provides instructions for making a protein called receptor expression-enhancing protein 1 (REEP1), which is found in nerve cells (neurons) in the brain and spinal cord. The REEP1 protein is located within cell compartments called mitochondria, which are the energy-producing centers in cells, and the endoplasmic reticulum, which helps with protein processing and transport.

The REEP1 protein plays a role in forming the network of tubules that make up the structure of the endoplasmic reticulum, regulating its size and determining how many proteins it can process. As part of its role in the endoplasmic reticulum, the REEP1 protein enhances the activity of certain other proteins called G protein-coupled receptors. These receptor proteins are eventually embedded within the outer membrane of cells, where they relay chemical signals from outside the cell to the interior of the cell.

The function of the REEP1 protein in the mitochondria is unknown.

### Health Conditions Related to Genetic Changes

[distal hereditary motor neuropathy, type V](#)

[spastic paraplegia type 31](#)

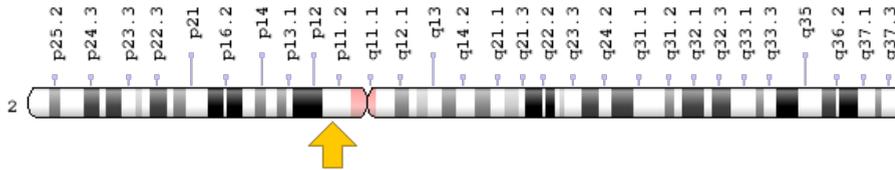
At least 44 mutations in the *REEP1* gene have been found to cause spastic paraplegia type 31. This condition is characterized by muscle stiffness (spasticity) and paralysis of the lower limbs (paraplegia) caused by degeneration of nerve cells (neurons) that trigger muscle movement. Most of the *REEP1* gene mutations that cause this condition insert or remove small pieces of DNA from the gene or alter the way the gene's instructions are used to make the protein. These mutations often result in a short, nonfunctional protein that is quickly broken down. As a result, there is a reduction in functional REEP1 protein.

It is unclear how *REEP1* gene mutations lead to the signs and symptoms of spastic paraplegia type 31. Researchers have shown that mitochondria in cells of affected individuals are less able to produce energy, which may contribute to the death of neurons and lead to the progressive movement problems of spastic paraplegia type 31; however, the exact mechanism that causes this condition is unknown.

## Chromosomal Location

Cytogenetic Location: 2p11.2, which is the short (p) arm of chromosome 2 at position 11.2

Molecular Location: base pairs 86,213,993 to 86,338,083 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- C2orf23
- FLJ13110
- receptor expression-enhancing protein 1
- REEP1\_HUMAN

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): G Protein-Coupled Receptors and Their Effectors  
<https://www.ncbi.nlm.nih.gov/books/NBK21718/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28REEP1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

### OMIM

- RECEPTOR EXPRESSION-ENHANCING PROTEIN 1  
<http://omim.org/entry/609139>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=REEP1%5Bgene%5D>
- HGNC Gene Family: Receptor accessory proteins  
<http://www.genenames.org/cgi-bin/genefamilies/set/717>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=25786](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=25786)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/65055>
- UniProt  
<http://www.uniprot.org/uniprot/Q9H902>

## **Sources for This Summary**

- Beetz C, Pieber TR, Hertel N, Schabhüttl M, Fischer C, Trajanoski S, Graf E, Keiner S, Kurth I, Wieland T, Varga RE, Timmerman V, Reilly MM, Strom TM, Auer-Grumbach M. Exome sequencing identifies a REEP1 mutation involved in distal hereditary motor neuropathy type V. *Am J Hum Genet.* 2012 Jul 13;91(1):139-45. doi: 10.1016/j.ajhg.2012.05.007. Epub 2012 Jun 14.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/22703882>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3397265/>
- Björk S, Hurt CM, Ho VK, Angelotti T. REEPs are membrane shaping adapter proteins that modulate specific g protein-coupled receptor trafficking by affecting ER cargo capacity. *PLoS One.* 2013 Oct 2;8(10):e76366. doi: 10.1371/journal.pone.0076366. eCollection 2013. Erratum in: *PLoS One.* 2013;8(12). doi:10.1371/annotation/6f86410c-63c3-4fcd-b1cb-9fd8d2ea95d0.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/24098485>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3788743/>
- Goizet C, Depienne C, Benard G, Boukhris A, Mundwiller E, Solé G, Coupry I, Pilliod J, Martin-Négrier ML, Fedirko E, Forlani S, Cazeneuve C, Hannequin D, Charles P, Feki I, Pinel JF, Ouvrard-Hernandez AM, Lyonnet S, Ollagnon-Roman E, Yaouanq J, Toutain A, Dussert C, Fontaine B, Leguern E, Lacombe D, Durr A, Rossignol R, Brice A, Stevanin G. REEP1 mutations in SPG31: frequency, mutational spectrum, and potential association with mitochondrial morpho-functional dysfunction. *Hum Mutat.* 2011 Oct;32(10):1118-27. doi: 10.1002/humu.21542. Epub 2011 Sep 9.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/21618648>
- Hurt CM, Björk S, Ho VK, Gilsbach R, Hein L, Angelotti T. REEP1 and REEP2 proteins are preferentially expressed in neuronal and neuronal-like exocytotic tissues. *Brain Res.* 2014 Jan 30;1545:12-22. doi: 10.1016/j.brainres.2013.12.008. Epub 2013 Dec 16.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/24355597>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3919455/>
- OMIM: RECEPTOR EXPRESSION-ENHANCING PROTEIN 1  
<http://omim.org/entry/609139>

- Schlang KJ, Arning L, Epplen JT, Stemmler S. Autosomal dominant hereditary spastic paraplegia: novel mutations in the REEP1 gene (SPG31). BMC Med Genet. 2008 Jul 21;9:71. doi: 10.1186/1471-2350-9-71.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/18644145>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2492855/>
  - Züchner S, Wang G, Tran-Viet KN, Nance MA, Gaskell PC, Vance JM, Ashley-Koch AE, Pericak-Vance MA. Mutations in the novel mitochondrial protein REEP1 cause hereditary spastic paraplegia type 31. Am J Hum Genet. 2006 Aug;79(2):365-9. Epub 2006 May 26.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/16826527>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1559498/>
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<https://ghr.nlm.nih.gov/gene/REEP1>

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